Dopa-responsive childhood dystonia: a forme fruste with writer's cramp, triggered by exercise

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Abstract

An 11-year-old girl was evaluated for walking difficulties and fatigue at the end of the day in the last 2 years. Handwriting was also difficult with ‘cramps’ after a short time of writing. Neurological examination was normal most of the time but in the evening and after exercise, an abnormal walking posture and rare dystonic movements of the foot could sometimes be seen. The mother was found to have mild parkinsonism and is asymptomatic on L-dopa. In the daughter, all symptoms and signs disappeared on L-dopa, but returned when the drug was withdrawn. The changes on- and off-treatment were documented with videofilms and computerized analysis of writing samples. The situation has been stable during a 5-year follow-up. We draw attention to this ‘forme fruste’ of dopa-sensitive childhood dystonia which becomes manifest with exercise and which can easily go unrecognized. We also discuss and illustrate the methods used for the analysis of writing.

Reference


Dopa-responsive childhood dystonia: a forme fruste with writer's cramp, triggered by exercise

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Here we report the case of a girl with this disorder diagnosed at the age of 11 years and followed for 5 years. Gait disturbances and dystonia were evident only in the evening and/or after exercise. She also had writer's cramp. All symptoms disappeared with L-dopa.

We wish to draw attention to the role of exercise in bringing about symptoms and to the importance of writer's cramp as a dystonic symptom (Sheehy and Marsden 1983; Deonna and Ferreira 1987), and also to illustrate the methods used to evaluate this latter problem and its response to treatment.

Case report
An 11-year-old left-handed girl was referred to our paediatric clinic from the orthopaedic department where she had been evaluated for foot deformity and a history of several 'ankle twisting' episodes. For the past 1 to 2 years, she had complained of fatigue and walking difficulties at the end of the day. She said that her foot suddenly twisted and that her legs rubbed each other. She could not wear regular shoes anymore, but only sport-type shoes. She was better in the morning or after prolonged rest. Despite these complaints, she did gymnastics and athletics. She also complained of tiring easily after a short period of writing and was in trouble with her teachers because of her slow and untidy written work.

On the first examination at 4 pm her gait was normal: on tiptoe walking however, she had a strange gait and rubbed her ankles against each other. She could walk on her heels normally. When lying supine, there was abnormal posture with
plantar flexion and inversion of the feet, more apparent on the left. No evident dystonic movements were seen. She wrote with a hyperextended wrist and stiff fingers and had to stop writing after a short period of time. Two days after this first visit, she was operated on for appendicitis, but had no symptoms when we saw her.

She was hospitalized 3 weeks later for a work-up. At this time (5pm) the examination was normal, except that her knees and feet occasionally rubbed each other while walking. The next morning, the neurological examination was entirely normal and none of the previous findings could be seen. Tiptoe walking and heel walking were normal again in the evening in the hospital. She was then asked to walk for half an hour and climb 15 flights of stairs in the hospital. After that, there was marked internal rotation of the thighs and plantar flexion of the feet when lying supine (not present in the morning and before exercise); during walking, the distance between the lower extremities had narrowed again. There was also definite intermittent dystonic posturing of the left foot during tiptoe walking.

"Writing posture and quality of written production deteriorated after 10 minutes (see details below). CT scan and nerve conduction velocities were normal."

A tentative diagnosis of DRD was made and she was started

Figure 1: Comparative writing samples: a) without L-dopa after 15 min writing; b) under L-dopa after identical duration of writing.

Figure 2a: Writing posture at start of writing

Figure 2b: Writing posture after 10 minutes of writing
on L-dopa 100mg + benzerazide 25mg twice a day for 4 days and increased to 3 times a day thereafter. All symptoms disappeared on this treatment.

Our patient is an only child. Her parents had no neurological complaints. One year after our diagnosis, the 43-year-old mother complained of progressive fatigue, slowness in her daily activities, a feeling of stiffness in her hands, difficulty with fast writing and some slowness in her gait. A diagnosis of mild parkinsonism was made. On L-dopa benzerazide (3 x 62.5mg), she showed definite improvement (knitting, writing, household tasks) and her neurological examination became normal. She did not wish to discontinue the medication over the next 2½ years.

Evaluation of the symptoms and signs on and off L-dopa
Because of the spontaneous fluctuations and subtlety of the neurological signs, and to try to confirm or disprove the subjective impression of cure while on treatment, the drug was stopped and restarted on several occasions and repeated video-filmed examinations were made in comparable situations (time of day, amount of physical effort). Symptoms reappeared each time after withdrawal and disappeared again on resumption of treatment.

The time for the symptoms to recur and their severity varied with the amount of physical activity or writing (of which there was a negligible amount during holidays). On a few occasions, they were much worse during intercurrent febrile illnesses.

The objective signs seen before treatment were again present off treatment at the end of the day and after a short time of writing.

Analysis of writing difficulties
The major subjective improvement was in handwriting and this made her life at school much easier. It also led to a marked improvement in school grades. To document this aspect, several parameters were studied.

Subjective description of the patient
She expressed the change as follows: ‘When I write, it is easier’: ‘I press less’, ‘I lean less with my hand’, ‘I get tired less quickly’, ‘I hold longer’.

Quality of written production
See Figure 1.

Writing speed
The same text after a comparable period of writing before the beginning of the test was on treatment, 6 minutes; off treatment, 7½ minutes.

Writing posture
Figure 2 shows the normal wrist posture on treatment and the abnormal hyperextended posture off treatment.

Computerized analysis of writing samples
One hundred and thirty samples produced by the patient (1) off treatment (24 June 1994) and (2) on treatment (27 February 1995) were recorded by means of a ZedPen and a digitizing tablet (sampling rate = 200Hz; spatial accuracy ±0.25mm). In each session the patient was asked to write words and simple geometrical patterns in several conditions (size, velocity and friction force variations) before and after an 18 minute exercise of continuous writing from dictation. Several spatial (trajectory length), temporal (duration), kinetic, static (average velocity and movement fluency) and dynamic (pen pressure) parameters were subsequently extracted from her samples (for description of material and method, see Teulings and Thomassen 1979, Teulings and Maarse 1984).

Spatial and temporal variability was evaluated by computing the relative length and the relative duration of strokes con-

Table 1: Quantitative data of writing parameters before and after 18min of writing exercise

<table>
<thead>
<tr>
<th>Parameter</th>
<th>Off L-dopa</th>
<th>On L-dopa</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Before</td>
<td>After</td>
</tr>
<tr>
<td>Spatial variability (SNR in arbitrary units)</td>
<td>11.9 6.7 18.0 7.2</td>
<td></td>
</tr>
<tr>
<td>Average pen pressure (g)</td>
<td>200–250 320–350 200–250 200–250</td>
<td></td>
</tr>
</tbody>
</table>
tained in the same word produced repetitively under different conditions. The signal-noise ratio (hereafter SNR; see Trudings and Schoumaker 1993) was then computed on these relative values. The signal represents intentional variations of the length or of the duration of strokes (e.g., the fact that the letter 'I' is about twice as high as the letter 'c'). The noise corresponds to random variations (e.g., size differences between various replications of the same letter): the lower the SNR, the more variable the production. Without treatment, the patient's writing before the 18-minute writing exercise was mainly characterized by a low value of the SNR computed on the relative stroke length (high spatial variability), as can be seen in Table 1. In comparison, neurologically normal, young adolescent females (12-year-olds) obtain scores averaging around 18 (Zesiger 1985). All other parameters, including pen pressure, fell within the normal range. After the writing exercise, there is a marked increase in spatial variability (lower SNR) and in pen pressure. Both values are clearly pathological: the patient's performance was poorer than that of an average 8-year-old child.

Under L-dopa, all parameters improved and were considered normal. Furthermore, no significant change was noted between writing samples recorded before and after the exercise. Samples of her writing and of their corresponding velocity and pressure profiles as a function of time are represented in Fig. 3. It can be seen that off treatment (top panel), letters were poorly formed and pen pressure is high. On treatment (bottom panel), letters were well formed and pen pressure was lower.

Discussion

Our case fits the category of dopamine-responsive dystonias (DRD). The diurnal fluctuation of symptoms and positive family history suggest a diagnosis of Segawa syndrome: hereditary progressive childhood dystonia (HPD). The mother has mild parkinsonism rather than dystonia as can be seen in older members of a family with DRD (Nygaard et al. 1992, Nygaard 1995). The recent genetic findings in HPD will probably help to determine whether there are different genetic entities within DRDs (Tanaka et al. 1995, Ichinose et al. 1994).

In any case, several particular clinical features of this case are important to recognize. First is the mild degree of involvement and absence of progression of the disease during the 5 years of observation. Gait and posture were entirely normal in the morning and only fatigue during writing was present. Although there is a wide variation in the severity and rate of progression of symptoms in dopamine-sensitive childhood dystonias (Deonna et al. 1986), such a form of dystonia has not been described.

Exertion clearly exacerbated the symptoms in our patient and intermittent foot dystonia was seen on some occasions only after heavy exercise. Worsening of the symptoms during the day, typical of the syndrome, is usually thought of as being related to the time since the last sleep rather than to exertion, although it is difficult to separate these aspects. The role of exertion itself might be more important than recognized and should be considered in suspected cases.

Dystonic writer's cramp, which had made our patient's school-life miserable, was the most disturbing symptom and was relieved by L-dopa.

Writer's cramp is thought to be less responsive to L-dopa than other dystonic symptoms, although it is difficult to judge objectively, and has not been studied in detail. We have shown how this very important aspect can be documented objectively by looking both qualitatively and quantitatively at different parameters characterizing the process as well as the product of writing. The technology used to measure several characteristics (spatial, temporal, and kinematic) can be a tool for following such situations. Although the normative data used in our study have been obtained in right-handers, it is known that the handwriting of left-handers is as fast and as efficient as that of right-handers. Both in adults (Wing 1980, Meunenbroek and Van Galen 1989) and in children (Zesiger 1984). Therefore, we do not believe that handedness was a confounding factor in our case.

Despite the benign course, mild and often occult symptoms of our patient's disease, her unexplained chronic evening fatigue, feet and writing problems led to several consultations and interpretations: they had a definite impact on her life and cannot be considered medically insignificant. Her condition could have remained totally unrecognized if the diagnosis had not been suspected by the referring physician and if she had not been seen first in the morning and without previous physical exercise. Incidentally, it was only by chance that the first consultation occurred in the evening and at an early phase of her appendicitis which made the signs more obvious than we ever saw them later.

Dopa-responsive dystonias are receiving considerable attention and major progress has been made at the recognition of the various facets of this condition particularly at the genetic level (Ichinose et al. 1994, Tanaka et al. 1995).

The clinical spectrum may still be expanding by the discovery of cases such as the one reported here. It remains to be seen whether there is a small subgroup among the numerous schoolchildren who suffer from writing problems (fatigue, slowness, cramps, poor writing) who have, in fact, a dystonic writer's cramp possibly responsive to L-dopa.

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